EARS2 gene

glutamyl-tRNA synthetase 2, mitochondrial

Normal Function

The *EARS2* gene provides instructions for making an enzyme called mitochondrial glutamyl-tRNA synthetase. This enzyme is important in the production (synthesis) of proteins in cellular structures called mitochondria, the energy-producing centers in cells. While most protein synthesis occurs in the fluid surrounding the cell nucleus (cytoplasm), some proteins are synthesized in the mitochondria.

During protein synthesis, in either the mitochondria or the cytoplasm, a type of RNA called transfer RNA (tRNA) helps assemble protein building blocks called amino acids into a chain that forms the protein. Each tRNA carries a specific amino acid to the growing chain. Enzymes called aminoacyl-tRNA synthetases, including mitochondrial glutamyl-tRNA synthetase, attach a particular amino acid to a specific tRNA. Mitochondrial glutamyl-tRNA synthetase attaches the amino acid glutamate to the correct tRNA, which helps ensure that glutamate is added at the proper place in the mitochondrial protein.

Health Conditions Related to Genetic Changes

Leigh syndrome

leukoencephalopathy with thalamus and brainstem involvement and high lactate

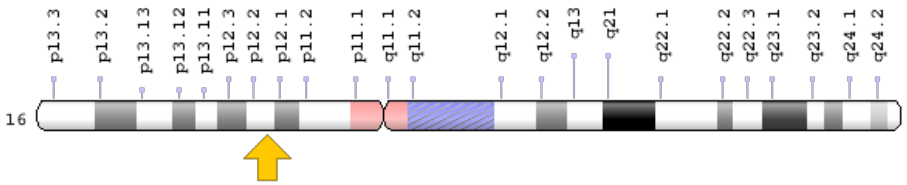
At least 23 mutations in the *EARS2* gene have been found in individuals with leukoencephalopathy with thalamus and brainstem involvement and high lactate (LTBL), a condition characterized by abnormalities in certain brain regions, including the thalamus and the brainstem (the part of the brain that connects to the spinal cord), and a high level of a substance called lactate in the brain and elsewhere in the body. Affected individuals typically have problems with thinking and motor abilities and with controlling muscle function.

The *EARS2* gene mutations involved in LTBL likely reduce the amount of mitochondrial glutamyl-tRNA synthetase. A shortage of this protein is thought to prevent the normal assembly of new proteins within mitochondria. Researchers speculate that impaired protein assembly disrupts mitochondrial energy production. However, it is unclear exactly how *EARS2* gene mutations lead to the features of LTBL.

Chromosomal Location

Cytogenetic Location: 16p12.2, which is the short (p) arm of chromosome 16 at position 12.2

Molecular Location: base pairs 23,522,013 to 23,557,375 on chromosome 16 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- COXPD12
- gluRS
- glutamate--tRNA ligase
- glutamate tRNA ligase 2, mitochondrial
- KIAA1970
- MSE1

Additional Information & Resources

Educational Resources

- Genomes (second edition, 2002: Aminoacyl-tRNA Synthetases Attach Amino Acids to tRNAs
  https://www.ncbi.nlm.nih.gov/books/NBK21111/#A7614

GeneReviews

- Leukodystrophy Overview
  https://www.ncbi.nlm.nih.gov/books/NBK184570

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28EARS2%5BTIAB%5D%29+AND+english%5Bla%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- GLUTAMYL-tRNA SYNTHETASE 2
  http://omim.org/entry/612799

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=EARS2%5Bgene%5D
- HGNC Gene Family: Aminoacyl tRNA synthetases, Class I
  http://www.genenames.org/cgi-bin/genefamilies/set/131
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=29419
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/124454
- UniProt
  http://www.uniprot.org/uniprot/Q5JPH6

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